



## spastic paraplegia type 7

Spastic paraplegia type 7 is part of a group of genetic disorders known as hereditary spastic paraplegias. These disorders are characterized by progressive muscle stiffness (spasticity) and the development of paralysis of the lower limbs (paraplegia). Hereditary spastic paraplegias are divided into two types: pure and complex. The pure types involve the lower limbs. The complex types involve the lower limbs and can also affect the upper limbs to a lesser degree; the structure or functioning of the brain; and the nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat, and sound (the peripheral nervous system). Spastic paraplegia type 7 can occur in either the pure or complex form.

Like all hereditary spastic paraplegias, spastic paraplegia type 7 involves spasticity of the leg muscles and increased muscle weakness. People with this form of spastic paraplegia can also experience exaggerated reflexes (hyperreflexia) in the arms; speech difficulties (dysarthria); difficulty swallowing (dysphagia); involuntary movements of the eyes (nystagmus); mild hearing loss; abnormal curvature of the spine (scoliosis); high-arched feet (pes cavus); numbness, tingling, or pain in the arms and legs (sensory neuropathy); disturbance in the nerves used for muscle movement (motor neuropathy); and muscle wasting (amyotrophy). The onset of symptoms varies greatly among those with spastic paraplegia type 7; however, abnormalities in muscle tone and other features are usually noticeable in adulthood.

### Frequency

The prevalence of all hereditary spastic paraplegias combined is estimated to be 2 to 6 in 100,000 people worldwide. Spastic paraplegia type 7 likely accounts for only a small percentage of all spastic paraplegia cases.

### Genetic Changes

Mutations in the *SPG7* gene cause spastic paraplegia type 7. The *SPG7* gene provides instructions for producing a protein called paraplegin. Located within the inner membrane of the energy-producing centers of cells (mitochondria), paraplegin is one of the proteins that form a complex called the m-AAA protease. The m-AAA protease is responsible for assembling ribosomes (cellular structures that process the cell's genetic instructions to create proteins) and removing nonfunctional proteins in the mitochondria. When there is a mutation in paraplegin, the m-AAA protease cannot function correctly. Nonfunctional m-AAA proteases cause a build up of unusable proteins in the mitochondria of nerve cells, which can result in swelling of the cell,

reduced cell signaling, and impaired cell movement, leading to the major signs and symptoms of spastic paraplegia type 7.

### **Inheritance Pattern**

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

### **Other Names for This Condition**

- Autosomal Recessive Hereditary Spastic Paraplegia
- Hereditary Spastic Paraplegia
- hereditary spastic paraplegia, paraplegin type
- spastic paraplegia 7

### **Diagnosis & Management**

These resources address the diagnosis or management of spastic paraplegia type 7:

- GeneReview: Hereditary Spastic Paraplegia Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1509>
- GeneReview: Spastic Paraplegia 7  
<https://www.ncbi.nlm.nih.gov/books/NBK1107>
- Genetic Testing Registry: Spastic paraplegia 7  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1846564/>
- Spastic Paraplegia Foundation, Inc.: Treatments and Therapies  
<http://sp-foundation.org/understanding-pls-hsp/treatments.html>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Neurologic Diseases  
<https://medlineplus.gov/neurologicdiseases.html>
- Health Topic: Neuromuscular Disorders  
<https://medlineplus.gov/neuromusculardisorders.html>
- Health Topic: Paralysis  
<https://medlineplus.gov/paralysis.html>
- Health Topic: Peripheral Nerve Disorders  
<https://medlineplus.gov/peripheralnervedisorders.html>

### Genetic and Rare Diseases Information Center

- Hereditary spastic paraplegia  
<https://rarediseases.info.nih.gov/diseases/6637/hereditary-spastic-paraplegia>

### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Hereditary Spastic Paraplegia  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Hereditary-spastic-paraplegia-Information-Page>

### Educational Resources

- Disease InfoSearch: Spastic paraplegia 7  
<http://www.diseaseinfosearch.org/Spastic+paraplegia+7/6699>
- MalaCards: spastic paraplegia 7  
[http://www.malacards.org/card/spastic\\_paraplegia\\_7](http://www.malacards.org/card/spastic_paraplegia_7)
- Merck Manual Consumer Version  
<http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/spinal-cord-disorders/hereditary-spastic-paraparesis>
- Orphanet: Hereditary spastic paraplegia  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=685](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=685)

### Patient Support and Advocacy Resources

- National Ataxia Foundation  
<http://www.ataxia.org/>
- National Organization for Rare Disorders (NORD): Hereditary Spastic Paraplegia  
<https://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/>

- RareConnect  
<https://www.rareconnect.org/en/community/hereditary-spastic-paraplegia>
- Spastic Paraplegia Foundation, Inc.: About Hereditary Spastic Paraplegia  
<http://sp-foundation.org/understanding-pls-hsp/hsp.html>

#### GeneReviews

- Hereditary Spastic Paraplegia Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1509>
- Spastic Paraplegia 7  
<https://www.ncbi.nlm.nih.gov/books/NBK1107>

#### Genetic Testing Registry

- Spastic paraplegia 7  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1846564/>

#### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22spastic+paraplegia+type+7%22+OR+%22Spastic+Paraplegia%2C+Hereditary%22+OR+%22Spastic+Paraplegia%22>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG7%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

#### OMIM

- SPASTIC PARAPLEGIA 7, AUTOSOMAL RECESSIVE  
<http://omim.org/entry/607259>

### **Sources for This Summary**

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